EHLERS-DANLOS SYNDROME– AN OVERVIEW

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Abstract

Ehlers-Danlos syndrome, hypermobility type is generally considered the least severe type of EDS, although significant complications, primarily musculoskeletal, can and do occur. The skin is often soft or velvety and may be mildly hyperextensible. Subluxations and dislocations are common; they may occur spontaneously or with minimal trauma and can be acutely painful. Degenerative joint disease is common. Chronic pain, distinct from that associated with acute dislocations or advanced osteoarthritis, is a serious complication of the condition and can be both physically and psychologically disabling. Easy bruising is common.

Keywords: Ehlers-Danlos Syndrome, Hypermobility, Management

1. Introduction:

Ehlers-Danlos syndrome (EDS), hypermobility type is generally considered the least severe type of EDS, although significant complications, primarily musculoskeletal, can and do occur. The skin is often soft or velvety and may be mildly hyperextensible. Subluxations and dislocations are common; they may occur spontaneously or with minimal trauma and can be acutely painful. Degenerative joint disease is common. Chronic pain, distinct from that associated with acute dislocations or advanced osteoarthritis, is a serious complication of the condition and can be both physically and psychologically disabling. Easy bruising is common. The diagnosis of EDS, hypermobility type is based entirely on clinical evaluation and family history. Some common features include:

• Passive dorsiflexion of each fifth finger greater than 90°
• Passive apposition of each thumb to the flexor surface of the forearm
• Hyperextension of each elbow greater than 10°
• Hyperextension of each knee greater than 10°
• Ability to place the palms on the floor with the knees fully extended
• Soft skin with normal or only slightly increased extensibility.
• Soft skin is subjectively assessed, preferably in an area in which moisturizer has not been applied.
• Skin hyperextensibility is assessed at a site lacking excess or loose skin and without evidence of prior trauma by gently pulling until resistance is met. An ideal location is the volar surface of the forearm, where the upper limit of normal extensibility is 1-1.5 cm. Extensor surfaces of joints have excess skin and should not be used.
• Spontaneous or easily induced skin cuts or tears
• Spontaneous or easily induced tears or ruptures of tendons, ligaments, vessels, or other internal organs
• Surgical complications, such as vessel rupture or sutures tearing through tissues and failing to hold
• Spontaneous wound dehiscence
• Recurrent or incisional hernias
• Significant skin hyperextensibility (>1.5 cm on the volar surface of the forearm)
• Thin, translucent skin

2. Clinical Diagnosis8–15:

Clinical diagnostic criteria and a revised nomenclature for all forms of Ehlers-Danlos syndrome (EDS) were proposed by Beighton . EDS, hypermobility type is characterized chiefly by joint laxity with soft skin and easy bruising, but other organ systems (especially gastrointestinal and cardiovascular) are frequently involved. It is distinguished from EDS, classic type by the more significant skin and soft tissue manifestations in the latter. The
Atrophic scars
Positive family history of EDS, hypermobility type (or family history of joint laxity), without significant skin or soft tissue fragility, in a pattern consistent with autosomal dominant inheritance
Recurrent joint dislocations or subluxations
Chronic joint, limb, and/or back pain
Easy bruising
Functional bowel disorders (functional gastritis, irritable bowel syndrome)
Neurally-mediated hypotension or postural orthostatic tachycardia
High, narrow palate
Dental crowding

3. Management:
- Treatment of manifestations: Physical therapy tailored to the individual; assistive devices (braces to improve joint stability; wheelchair or scooter to offload stress on lower-extremity joints; suitable mattress to improve sleep quality); pain medication tailored to symptoms; appropriate therapy for gastritis/reflux /delayed gastric emptying/irritable bowel syndrome; possible beta-blockade for progressive aortic enlargement; psychological and/or pain-oriented counseling.
- Prevention of primary manifestations: Low-resistance exercise to increase muscle tone for improved joint stability; appropriate writing utensils to reduce finger and hand strain.
- Prevention of secondary complications: Calcium, vitamin D, low-impact weight-bearing exercise to maximize bone density.
- Surveillance: DEXA every other year if bone loss is confirmed.
- Agents/circumstances to avoid: Joint hyperextension; resistance/isometric exercise can exacerbate joint instability and pain; high-impact activity increases the risk of acute subluxation/dislocation, chronic pain, and osteoarthritis; cautious use of crutches, canes, and walkers, which put increased stress on the upper extremities.
- Genetic counseling: EDS, hypermobility type is inherited in an autosomal dominant manner. Most individuals diagnosed with the syndrome have an affected parent. The proportion of cases caused by de novo mutations is unknown. Each child of an individual with EDS, hypermobility type has a 50% chance of inheriting the disorder. Prenatal testing is not available.

4. Testing: Different types of testing are as follows:
The biochemical etiology of EDS, hypermobility type is unknown in most cases.
- Molecular Genetic Testing: Haploinsufficiency of tenascin X, encoded by TNXB, has been associated with EDS, hypermobility type in a small subset of individuals. Haploinsufficiency of tenascin X appears to confer typical joint manifestations and soft skin, without skin hyperextensibility or hematologic manifestations.
- Other loci. The etiology and genetic locus (or loci) are unknown in the vast majority of cases.
- Clinical testing. Sequence analysis of TNXB is available clinically on a limited basis.
- Research testing. Serum tenascin X protein testing is available on a research basis only.

5. Testing Strategy:
- To confirm/establish the diagnosis in a proband: No clinical diagnostic testing is indicated to confirm or establish the diagnosis in a proband. TNXB sequencing could be considered in persons with joint laxity in the absence of easy bruising or skin hyperextensibility.
- Genetically Related Disorders: An autosomal recessive form of Ehlers-Danlos syndrome has been described in individuals with tenascin-X deficiency resulting from compound heterozygous or homozygous TNXB mutation/deletion [Bristow et al 2005]. Clinical features include joint laxity, hyper-extensible skin, and easy bruising with normal wound healing and absence of atrophic scarring. Some, but not all, also have congenital adrenal hyperplasia as a result of contiguous gene deletion involving CYP21A2.

References:


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